

## SEQUENCE LISTING

IAP13 Rec'd PCT/PTO 09 DEC 2005

<110> Pietrangelo, Antonello  
 <120> Mutations in the SLC40A1 gene associated to impaired iron homeostasis  
 <130> 8907-109-999  
 <140> To be Determined  
 <141> 2005-12-09  
 <150> PCT/EP2004/051068  
 <151> 2004-06-09  
 <160> 30  
 <170> PatentIn version 3.1  
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 <212> DNA  
 <213> Homo sapiens  
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 <222> (1)..(1716)  
 <223> cDNA encoding wild type ferroportin 1. Polymorphisms related to the codons:  
 238-240 (G80), 520-522 (N174), 742-744 (Q248)  
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 Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val  
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 Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
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 ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc ggt 240  
 Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly  
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 Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu  
 85 90 95  
 gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg 336  
 Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met  
 100 105 110  
 gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt 384

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Glu	Glu	Glu	Thr	Glu	Leu	Lys	Gln	Leu	Asn	Leu	His	Lys	Asp	Thr	Glu		
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cca	aaa	ccc	ctg	gag	gga	act	cat	cta	atg	ggt	gtg	aaa	gac	tct	aac	816	
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Ile	His	Glu	Leu	Glu	His	Glu	Gln	Glu	Pro	Thr	Cys	Ala	Ser	Gln	Met		
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Ala	Glu	Pro	Phe	Arg	Thr	Phe	Arg	Asp	Gly	Trp	Val	Ser	Tyr	Tyr	Asn		
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tca att aca cct acc aag ata cct gaa att aca act gaa ata tac atg Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met 420 425 430			1296
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tct gtg ccc ata atc tct gtc agt ctg ctg ttt gca ggc gtc att gct Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala 450 455 460			1392
gct aga atc ggt ctt tgg tcc ttt gat tta act gtg aca cag ttg ctg Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu 465 470 475 480			1440
caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln 485 490 495			1488
aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile 500 505 510			1536
ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val 515 520 525			1584
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 <213> Homo sapiens

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 35 40 45  
 Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
 50 55 60  
 Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly  
 65 70 75 80  
 Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu  
 85 90 95  
 Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met  
 100 105 110  
 Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val  
 115 120 125  
 Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn  
 130 135 140  
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 145 150 155 160  
 Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr  
 165 170 175  
 Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val  
 180 185 190  
 Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile  
 195 200 205  
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 210 215 220  
 Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys  
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Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu  
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 Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn  
 260 265 270  
 Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met  
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 Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn  
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 Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr  
 305 310 315 320  
 Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly  
 325 330 335  
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 Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly  
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 Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu  
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 385 390 395 400  
 Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu  
 405 410 415  
 Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met  
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 Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu  
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 Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala  
 450 455 460  
 Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu  
 465 470 475 480  
 Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln

	485		490		495
Asn Ser Met	Asn Tyr Leu Leu Asp	Leu Leu His Phe Ile Met Val Ile			
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Leu Ala Pro	Asn Pro Glu Ala Phe Gly Leu Leu Val	Leu Ile Ser Val			
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Ser Phe Val	Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn				
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 <223> cDNA encoding a ferroportin 1 mutated in position (G80).

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tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg	144
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val	
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Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr	
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ata	cga	agg	att	gac	cag	tta	acc	aac	atc	tta	gcc	ccc	atg	gct	gtt	576		
Ile	Arg	Arg	Ile	Asp	Gln	Leu	Thr	Asn	Ile	Leu	Ala	Pro	Met	Ala	Val			
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Gly	Gln	Ile	Met	Thr	Phe	Gly	Ser	Pro	Val	Ile	Gly	Cys	Gly	Phe	Ile			
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tcg	gga	tgg	aac	ttg	gta	tcc	atg	tgc	gtg	gag	tac	gtc	ctg	ctc	tgg	672		
Ser	Gly	Trp	Asn	Leu	Val	Ser	Met	Cys	Val	Glu	Tyr	Val	Leu	Leu	Trp			
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Lys	Val	Tyr	Gln	Lys	Thr	Pro	Ala	Leu	Ala	Val	Lys	Ala	Gly	Leu	Lys			
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gaa	gag	gaa	act	gaa	ttg	aaa	cag	ctg	aat	tta	cac	aaa	gat	act	gag	768		
Glu	Glu	Glu	Thr	Glu	Leu	Lys	Gln	Leu	Asn	Leu	His	Lys	Asp	Thr	Glu			
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cca	aaa	ccc	ctg	gag	gga	act	cat	cta	atg	ggg	gtg	aaa	gac	tct	aac	816		
Pro	Lys	Pro	Leu	Glu	Gly	Thr	His	Leu	Met	Gly	Val	Lys	Asp	Ser	Asn			
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Val	Leu	Gly	Phe	Asp	Cys	Ile	Thr	Thr	Gly	Tyr	Ala	Tyr	Thr	Gln	Gly			
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ctg	agt	ggg	tcc	atc	ctc	agt	att	ttg	atg	gga	gca	tca	gct	ata	act	1056		
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Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu	
370 375 380	
atc ttg tgt gtg atc tct gta ttc atg cct gga agc ccc ctg gac ttg	1200
Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu	
385 390 395 400	
tcc gtt tct cct ttt gaa gat atc cga tca agg ttc att caa gga gag	1248
Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu	
405 410 415	
tca att aca cct acc aag ata cct gaa att aca act gaa ata tac atg	1296
Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met	
420 425 430	
tct aat ggg tct aat tct gct aat att gtc ccg gag aca agt cct gaa	1344
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435 440 445	
tct gtg ccc ata atc tct gtc agt ctg ctg ttt gca ggc gtc att gct	1392
Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala	
450 455 460	
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Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu	
465 470 475 480	
caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag	1488
Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln	
485 490 495	
aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc	1536
Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile	
500 505 510	
ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc	1584
Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val	
515 520 525	
tcc ttt gtg gca atg ggc cac att atg tat ttc cga ttt gcc caa aat	1632
Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn	
530 535 540	
act ctg gga aac aag ctc ttt gct tgc ggt cct gat gca aaa gaa gtt	1680
Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val	
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agg aag gaa aat caa gca aat aca tct gtt gtt tga	1716
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 <212> PRT



<213> Homo sapiens

<400> 4

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35 40 45

Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
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Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Ser  
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Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu  
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Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met  
100 105 110

Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val  
115 120 125

Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn  
130 135 140

Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val  
145 150 155 160

Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr  
165 170 175

Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val  
180 185 190

Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile  
195 200 205

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210 215 220

Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys

225		230		235		240									
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Pro	Lys	Pro	Leu	Glu	Gly	Thr	His	Leu	Met	Gly	Val	Lys	Asp	Ser	Asn
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Ile	His	Glu	Leu	Glu	His	Glu	Gln	Glu	Pro	Thr	Cys	Ala	Ser	Gln	Met
		275					280					285			
Ala	Glu	Pro	Phe	Arg	Thr	Phe	Arg	Asp	Gly	Trp	Val	Ser	Tyr	Tyr	Asn
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Gln	Pro	Val	Phe	Leu	Ala	Gly	Met	Gly	Leu	Ala	Phe	Leu	Tyr	Met	Thr
305					310					315					320
Val	Leu	Gly	Phe	Asp	Cys	Ile	Thr	Thr	Gly	Tyr	Ala	Tyr	Thr	Gln	Gly
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Leu	Ser	Gly	Ser	Ile	Leu	Ser	Ile	Leu	Met	Gly	Ala	Ser	Ala	Ile	Thr
			340					345					350		
Gly	Ile	Met	Gly	Thr	Val	Ala	Phe	Thr	Trp	Leu	Arg	Arg	Lys	Cys	Gly
		355					360					365			
Leu	Val	Arg	Thr	Gly	Leu	Ile	Ser	Gly	Leu	Ala	Gln	Leu	Ser	Cys	Leu
	370					375					380				
Ile	Leu	Cys	Val	Ile	Ser	Val	Phe	Met	Pro	Gly	Ser	Pro	Leu	Asp	Leu
385					390					395					400
Ser	Val	Ser	Pro	Phe	Glu	Asp	Ile	Arg	Ser	Arg	Phe	Ile	Gln	Gly	Glu
				405					410					415	
Ser	Ile	Thr	Pro	Thr	Lys	Ile	Pro	Glu	Ile	Thr	Thr	Glu	Ile	Tyr	Met
			420					425					430		
Ser	Asn	Gly	Ser	Asn	Ser	Ala	Asn	Ile	Val	Pro	Glu	Thr	Ser	Pro	Glu
		435					440					445			
Ser	Val	Pro	Ile	Ile	Ser	Val	Ser	Leu	Leu	Phe	Ala	Gly	Val	Ile	Ala
	450					455					460				
Ala	Arg	Ile	Gly	Leu	Trp	Ser	Phe	Asp	Leu	Thr	Val	Thr	Gln	Leu	Leu
465					470					475					480

Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln  
485 490 495

Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile  
500 505 510

Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val  
515 520 525

Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn  
530 535 540

Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val  
545 550 555 560

Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val  
565 570

<210> 5

<211> 1716

<212> DNA

<213> Homo sapiens

<220>

<221> CDS

<222> (1)..(1716)

<223> cDNAencoding a ferroportin 1 mutated in position 174 (N174)

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Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser  
1 5 10 15

ttg gcc gac tac ctg acc tct gca aaa ttc ctt ctc tac ctt ggt cat 96  
Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His  
20 25 30

tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg 144  
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val  
35 40 45

ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac 192  
Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
50 55 60

ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc ggt 240  
Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly  
65 70 75 80

gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg 288  
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu  
85 90 95

gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg	336
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met	
100 105 110	
gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt	384
Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val	
115 120 125	
ctc act tcc tgc tat atc ctg atc atc act att gca aat att gca aat	432
Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn	
130 135 140	
ttg gcc agt act gct act gca atc aca atc caa agg gat tgg att gtt	480
Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val	
145 150 155 160	
gtt gtt gca gga gaa gac aga agc aaa cta gca aat atg att gcc aca	528
Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Ile Ala Thr	
165 170 175	
ata cga agg att gac cag tta acc aac atc tta gcc ccc atg gct gtt	576
Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val	
180 185 190	
ggc cag att atg aca ttt ggc tcc cca gtc atc ggc tgt ggc ttt att	624
Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile	
195 200 205	
tcg gga tgg aac ttg gta tcc atg tgc gtg gag tac gtc ctg ctc tgg	672
Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp	
210 215 220	
aag gtt tac cag aaa acc cca gct cta gct gtg aaa gct ggt ctt aaa	720
Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys	
225 230 235 240	
gaa gag gaa act gaa ttg aaa cag ctg aat tta cac aaa gat act gag	768
Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu	
245 250 255	
cca aaa ccc ctg gag gga act cat cta atg ggt gtg aaa gac tct aac	816
Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn	
260 265 270	
atc cat gag ctt gaa cat gag caa gag cct act tgt gcc tcc cag atg	864
Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met	
275 280 285	
gct gag ccc ttc cgt acc ttc cga gat gga tgg gtc tcc tac tac aac	912
Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn	
290 295 300	
cag cct gtg ttt ctg gct ggc atg ggt ctt gct ttc ctt tat atg act	960
Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr	
305 310 315 320	
gtc ctg ggc ttt gac tgc atc acc aca ggg tac gcc tac act cag gga	1008
Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly	
325 330 335	

ctg agt ggt tcc atc ctc agt att ttg atg gga gca tca gct ata act	1056
Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr	
340 345 350	
gga ata atg gga act gta gct ttt act tgg cta cgt cga aaa tgt ggt	1104
Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly	
355 360 365	
ttg gtt cgg aca ggt ctg atc tca gga ttg gca cag ctt tcc tgt ttg	1152
Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu	
370 375 380	
atc ttg tgt gtg atc tct gta ttc atg cct gga agc ccc ctg gac ttg	1200
Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu	
385 390 395 400	
tcc gtt tct cct ttt gaa gat atc cga tca agg ttc att caa gga gag	1248
Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu	
405 410 415	
tca att aca cct acc aag ata cct gaa att aca act gaa ata tac atg	1296
Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met	
420 425 430	
tct aat ggg tct aat tct gct aat att gtc ccg gag aca agt cct gaa	1344
Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu	
435 440 445	
tct gtg ccc ata atc tct gtc agt ctg ctg ttt gca ggc gtc att gct	1392
Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala	
450 455 460	
gct aga atc ggt ctt tgg tcc ttt gat tta act gtg aca cag ttg ctg	1440
Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu	
465 470 475 480	
caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag	1488
Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln	
485 490 495	
aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc	1536
Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile	
500 505 510	
ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc	1584
Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val	
515 520 525	
tcc ttt gtg gca atg ggc cac att atg tat ttc cga ttt gcc caa aat	1632
Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn	
530 535 540	
act ctg gga aac aag ctc ttt gct tgc ggt cct gat gca aaa gaa gtt	1680
Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val	
545 550 555 560	
agg aag gaa aat caa gca aat aca tct gtt gtt tga	1716
Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val	
565 570	

<210> 6  
 <211> 571  
 <212> PRT  
 <213> Homo sapiens

<400> 6

Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser  
 1 5 10 15

Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His  
 20 25 30

Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val  
 35 40 45

Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
 50 55 60

Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly  
 65 70 75 80

Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu  
 85 90 95

Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met  
 100 105 110

Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val  
 115 120 125

Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn  
 130 135 140

Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val  
 145 150 155 160

Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Ile Ala Thr  
 165 170 175

Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val  
 180 185 190

Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile  
 195 200 205

Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp  
 210 215 220

Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys  
 225 230 235 240

Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu  
 245 250 255

Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn  
 260 265 270

Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met  
 275 280 285

Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn  
 290 295 300

Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr  
 305 310 315 320

Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly  
 325 330 335

Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr  
 340 345 350

Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly  
 355 360 365

Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu  
 370 375 380

Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu  
 385 390 395 400

Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu  
 405 410 415

Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met  
 420 425 430

Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu  
 435 440 445

Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala  
 450 455 460

Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu  
465 470 475 480

Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln  
485 490 495

Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile  
500 505 510

Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val  
515 520 525

Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn  
530 535 540

Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val  
545 550 555 560

Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val  
565 570

<210> 7  
<211> 1716  
<212> DNA  
<213> Homo sapiens

<220>  
<221> CDS  
<222> (1)..(1716)  
<223> cDNA encoding a ferroportina 1 mutated in position 248 (Q248).

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Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser  
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ttg gcc gac tac ctg acc tct gca aaa ttc ctt ctc tac ctt ggt cat 96  
Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His  
20 25 30  
tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg 144  
Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val  
35 40 45  
ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac 192  
Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
50 55 60  
ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc ggt 240  
Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly  
65 70 75 80



gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg	288
Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu	
85 90 95	
gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg	336
Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met	
100 105 110	
gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt	384
Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val	
115 120 125	
ctc act tcc tgc tat atc ctg atc atc act att gca aat att gca aat	432
Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn	
130 135 140	
ttg gcc agt act gct act gca atc aca atc caa agg gat tgg att gtt	480
Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val	
145 150 155 160	
gtt gtt gca gga gaa gac aga agc aaa cta gca aat atg aat gcc aca	528
Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr	
165 170 175	
ata cga agg att gac cag tta acc aac atc tta gcc ccc atg gct gtt	576
Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val	
180 185 190	
ggc cag att atg aca ttt ggc tcc cca gtc atc ggc tgt ggc ttt att	624
Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile	
195 200 205	
tcg gga tgg aac ttg gta tcc atg tgc gtg gag tac gtc ctg ctc tgg	672
Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp	
210 215 220	
aag gtt tac cag aaa acc cca gct cta gct gtg aaa gct ggt ctt aaa	720
Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys	
225 230 235 240	
gaa gag gaa act gaa ttg aaa cat ctg aat tta cac aaa gat act gag	768
Glu Glu Glu Thr Glu Leu Lys His Leu Asn Leu His Lys Asp Thr Glu	
245 250 255	
cca aaa ccc ctg gag gga act cat cta atg ggt gtg aaa gac tct aac	816
Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn	
260 265 270	
atc cat gag ctt gaa cat gag caa gag cct act tgt gcc tcc cag atg	864
Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met	
275 280 285	
gct gag ccc ttc cgt acc ttc cga gat gga tgg gtc tcc tac tac aac	912
Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn	
290 295 300	
cag cct gtg ttt ctg gct ggc atg ggt ctt gct ttc ctt tat atg act	960
Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr	
305 310 315 320	
gtc ctg ggc ttt gac tgc atc acc aca ggg tac gcc tac act cag gga	1008

Val	Leu	Gly	Phe	Asp 325	Cys	Ile	Thr	Thr	Gly 330	Tyr	Ala	Tyr	Thr	Gln 335	Gly		
ctg	agt	ggg	tcc	atc	ctc	agt	att	ttg	atg	gga	gca	tca	gct	ata	act		1056
Leu	Ser	Gly	Ser	Ile	Leu	Ser	Ile	Leu	Met	Gly	Ala	Ser	Ala	Ile	Thr		
			340					345					350				
gga	ata	atg	gga	act	gta	gct	ttt	act	tgg	cta	cgt	cga	aaa	tgt	ggg		1104
Gly	Ile	Met	Gly	Thr	Val	Ala	Phe	Thr	Trp	Leu	Arg	Arg	Lys	Cys	Gly		
		355					360					365					
ttg	gtt	cgg	aca	ggg	ctg	atc	tca	gga	ttg	gca	cag	ctt	tcc	tgt	ttg		1152
Leu	Val	Arg	Thr	Gly	Leu	Ile	Ser	Gly	Leu	Ala	Gln	Leu	Ser	Cys	Leu		
	370					375					380						
atc	ttg	tgt	gtg	atc	tct	gta	ttc	atg	cct	gga	agc	ccc	ctg	gac	ttg		1200
Ile	Leu	Cys	Val	Ile	Ser	Val	Phe	Met	Pro	Gly	Ser	Pro	Leu	Asp	Leu		
385					390					395					400		
tcc	gtt	tct	cct	ttt	gaa	gat	atc	cga	tca	agg	ttc	att	caa	gga	gag		1248
Ser	Val	Ser	Pro	Phe	Glu	Asp	Ile	Arg	Ser	Arg	Phe	Ile	Gln	Gly	Glu		
				405					410					415			
tca	att	aca	cct	acc	aag	ata	cct	gaa	att	aca	act	gaa	ata	tac	atg		1296
Ser	Ile	Thr	Pro	Thr	Lys	Ile	Pro	Glu	Ile	Thr	Thr	Glu	Ile	Tyr	Met		
			420					425					430				
tct	aat	ggg	tct	aat	tct	gct	aat	att	gtc	ccg	gag	aca	agt	cct	gaa		1344
Ser	Asn	Gly	Ser	Asn	Ser	Ala	Asn	Ile	Val	Pro	Glu	Thr	Ser	Pro	Glu		
		435					440					445					
tct	gtg	ccc	ata	atc	tct	gtc	agt	ctg	ctg	ttt	gca	ggc	gtc	att	gct		1392
Ser	Val	Pro	Ile	Ile	Ser	Val	Ser	Leu	Leu	Phe	Ala	Gly	Val	Ile	Ala		
	450					455					460						
gct	aga	atc	ggg	ctt	tgg	tcc	ttt	gat	tta	act	gtg	aca	cag	ttg	ctg		1440
Ala	Arg	Ile	Gly	Leu	Trp	Ser	Phe	Asp	Leu	Thr	Val	Thr	Gln	Leu	Leu		
465					470				475						480		
caa	gaa	aat	gta	att	gaa	tct	gaa	aga	ggc	att	ata	aat	ggg	gta	cag		1488
Gln	Glu	Asn	Val	Ile	Glu	Ser	Glu	Arg	Gly	Ile	Ile	Asn	Gly	Val	Gln		
			485						490					495			
aac	tcc	atg	aac	tat	ctt	ctt	gat	ctt	ctg	cat	ttc	atc	atg	gtc	atc		1536
Asn	Ser	Met	Asn	Tyr	Leu	Leu	Asp	Leu	Leu	His	Phe	Ile	Met	Val	Ile		
			500					505					510				
ctg	gct	cca	aat	cct	gaa	gct	ttt	ggc	ttg	ctc	gta	ttg	att	tca	gtc		1584
Leu	Ala	Pro	Asn	Pro	Glu	Ala	Phe	Gly	Leu	Leu	Val	Leu	Ile	Ser	Val		
		515					520					525					
tcc	ttt	gtg	gca	atg	ggc	cac	att	atg	tat	ttc	cga	ttt	gcc	caa	aat		1632
Ser	Phe	Val	Ala	Met	Gly	His	Ile	Met	Tyr	Phe	Arg	Phe	Ala	Gln	Asn		
	530					535					540						
act	ctg	gga	aac	aag	ctc	ttt	gct	tgc	ggg	cct	gat	gca	aaa	gaa	gtt		1680
Thr	Leu	Gly	Asn	Lys	Leu	Phe	Ala	Cys	Gly	Pro	Asp	Ala	Lys	Glu	Val		
545					550				555						560		
agg	aag	gaa	aat	caa	gca	aat	aca	tct	gtt	gtt	tga						1716
Arg	Lys	Glu	Asn	Gln	Ala	Asn	Thr	Ser	Val	Val							

565

570

&lt;210&gt; 8

&lt;211&gt; 571

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;400&gt; 8

Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser  
 1 5 10 15

Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His  
 20 25 30

Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val  
 35 40 45

Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr  
 50 55 60

Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly  
 65 70 75 80

Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu  
 85 90 95

Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met  
 100 105 110

Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val  
 115 120 125

Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn  
 130 135 140

Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val  
 145 150 155 160

Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr  
 165 170 175

Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val  
 180 185 190

Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile  
 195 200 205

Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp  
 210 215 220

Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys  
 225 230 235 240

Glu Glu Glu Thr Glu Leu Lys His Leu Asn Leu His Lys Asp Thr Glu  
 245 250 255

Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn  
 260 265 270

Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met  
 275 280 285

Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn  
 290 295 300

Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr  
 305 310 315 320

Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly  
 325 330 335

Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr  
 340 345 350

Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly  
 355 360 365

Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu  
 370 375 380

Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu  
 385 390 395 400

Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu  
 405 410 415

Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met  
 420 425 430

Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu  
 435 440 445

Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala  
 450 455 460

Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu  
 465 470 475 480

Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln  
 485 490 495

Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile  
 500 505 510

Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val  
 515 520 525

Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn  
 530 535 540

Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val  
 545 550 555 560

Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val  
 565 570

<210> 9  
 <211> 20  
 <212> DNA  
 <213> Homo sapiens

<220>  
 <221> polymerase chain reaction primer  
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 <223> 5' PCR primer. Exon 1

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20

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<220>  
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 <223> 3' PCR primer. Exon1

<400> 10  
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20

<210> 11  
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 <223> 5' PCR primer. Exon 2

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25

<210> 12  
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 <223> 3' PCR primer. exon 2

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24

<210> 13  
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 <223> 5' PCR primer. Exon 3

<400> 13  
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23

<210> 14  
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 <220>  
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 <223> 3' PCR primer. Exon 3

<400> 14  
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22

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<400> 15  
gagacatttt gatgtaatgt acac

24

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24

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<400> 17  
ccaccaaaga ctatttttaa ctgc

24

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<222> (1)..(24)  
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<400> 18  
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24

<210> 19

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 <210> 20  
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 <212> DNA  
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 cccactggt aataaaacct g 21  
  
 <210> 21  
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 <210> 23  
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 <223> 3' PCR primer. Exon 8  
  
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 gctgacttag gtttctctaaa cagc 24  
  
 <210> 25  
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 <212> DNA  
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 atcagtgact 10  
  
 <210> 26  
 <211> 10  
 <212> DNA  
 <213> Homo sapiens  
  
 <220>  
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 <223> oligonucleotide comprising the polymorphism at nt 521.  
  
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 <210> 27  
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<220>  
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10

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<220>  
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<223> X different from glycine

<400> 28

Ile Ile Xaa Asp Trp  
1 5

<210> 29  
<211> 5  
<212> PRT  
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<220>  
<221> MISC\_FEATURE  
<222> (1)..(5)  
<223> X different form asparagine

<400> 29

Asn Met Xaa Ala Thr  
1 5

<210> 30  
<211> 5  
<212> PRT  
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<220>  
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<222> (1)..(5)  
<223> X different from glutamine

<400> 30

Leu Lys Xaa Leu Asn  
1 5